

ABSTRACT

A method for detecting Paget disease of bone and an animal showing the pathology of Paget disease of bone are provided. In particular, there are provided associating
5 Paget disease of bone with the mutation a chondroitin/chondroitin synthase gene comprising the amino acid sequence depicted in any one of SEQ ID NO: 2, SEQ ID NO: 4, SEQ ID NO: 6, SEQ ID NO: 66, SEQ ID NO: 68 or SEQ ID
10 NO: 70 or the amount of expression of said gene, detection of Paget disease of bone, and a knockout animal prepared by knocking out the chondroitin/chondroitin synthase gene.